

Note: The following information is provided by the author(s) and has not been reviewed by GeneReviews staff.

Table 3. Mutations Found in Ataxia with Vitamin E Deficiency

| Exon/ Intron | Mutation | NT Position | NT Change | Predicted Effect on ATTP | References |
|-----------------|--------------------------------|----------------|-------------|---|---|
| 5'UTR | Kocak consensus sequence | -1 | C>T | No initiation of translation | Usuki & Maruyama 2000 |
| Exon 1 | M1T | 2 | ATG>ACG | Aberrant initiation of translation | Hoshino et al 1999 |
| Exon 1 | M1T | 2 | ATG>AGG | Aberrant initiation of translation | Schuelke, unpublished |
| Exon 1 | R59W | 175 | CGG>TGG | Missense | Cavalier et al 1998 |
| Exon 1 | D64G | 191 | GAC>GGC | Missense | Usuki and Maruyama 2000 |
| Intron 1 | Splice acceptor | IVS1-1 | G>C | Skipping of exon 2, frameshift after R68 | Cavalier et al 1998 |
| Exon 2 | Frameshift mutation | 219-220 | insAT | Frameshift after Y73 | Mariotti et al 2004 |
| Exon 2 | Frameshift mutation | 302-309 | delATGGAGTC | Y100 + 27 nonsense AA, then stop | Schuelke, unpublished |
| Exon 2 | H101Q | 303 | CAT>CAG | Missense | Gotoda et al 1995, Yokota et al 1996, Cavalier et al 1998 |
| Exon 2 | Splice-site mutation | 306 | GGA>GGG | Alternative splice-site activation | Cavalier et al 1998 |
| Exon 2 | A120T | 358 | GCA>ACA | Missense | Cavalier et al 1998 |
| Intron 2 | Splice donor | IVS2+1 | G>A | Skipping of exon 2, frameshift | Schuelke, unpublished |

| Exon/ Intron | Mutation | NT Position | NT Change | Predicted Effect on ATTP | References |
|-------------------------|---------------------|------------------------|------------------|---|---|
| | | | | after R68 | |
| Exon 3 | R134X | 400 | CGA>TGA | Nonsense | Cavalier et al 1998 |
| Exon 3 | E141K | 421 | GAG>AAG | Missense | Cavalier et al 1998; Schuelke, Elsner et al 2000 |
| Exon 3 | Frameshift mutation | 486 | del T | G162 + 12 nonsense AA, then stop | Hentati et al 1996, Cavalier et al 1998 |
| Exon 3 | Frameshift mutation | 513-514 | ins TT | I171 + 4 nonsense AA, then stop | Ouahchi et al 1995; Hentati et al 1996; Cavalier et al 1998; Schuelke, Elsner et al 2000 |
| Exon 3 | Frameshift mutation | 530-531 | AG>GTAAGT | T177 + 2 nonsense AA, then stop | Ouahchi et al 1995, Cavalier et al 1998 |
| Exon 3 | Splice donor | 552 | ACG>ACA | Skipping of exon 3, frameshift after I119 | Tamaru et al 1997, Schuelke et al 1999 |
| Exon 4 | L183P | 548 | CTT>CCT | Missense | Shimohata et al 1998 |
| Exon 4 | R192H | 575 | TTG>TCG | Missense | Hentati et al 1996, Cavalier et al 1998 |
| Exon 4 | R221W | 661 | CGG>TGG | Missense | Cavalier et al 1998 |
| Exon 5 | G246R | 736 | GGT>CGT | Missense | Mariotti et al 2004 |
| Exon 5 | Frameshift mutation | 744 | del A | E248 + 14 nonsense AA, then stop | Ouahchi et al 1995, Cavalier et al 1998 |